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List of Publications by Year
in descending order

508 papers	53,075 citations	1299 109 h-index	1796 211 g-index
528 all docs	528 docs citations	528 times ranked	32214 citing authors

#	Article	IF	Citations
1	Revised terminology and concepts for organization of seizures and epilepsies: Report of the ILAE Commission on Classification and Terminology, 2005â€“2009. Epilepsia, 2010, 51, 676-685.	2.6	3,612
2	<scp>ILAE</scp> classification of the epilepsies: Position paper of the <scp>ILAE</scp> Commission for Classification and Terminology. Epilepsia, 2017, 58, 512-521.	2.6	3,464
3	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	13.7	1,351
4	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
5	A missense mutation in the neuronal nicotinic acetylcholine receptor Î±4 subunit is associated with autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 1995, 11, 201-203.	9.4	1,074
6	A Potassium Channel Mutation in Neonatal Human Epilepsy. Science, 1998, 279, 403-406.	6.0	1,013
7	doublecortin, a Brain-Specific Gene Mutated in Human X-Linked Lissencephaly and Double Cortex Syndrome, Encodes a Putative Signaling Protein. Cell, 1998, 92, 63-72.	13.5	1,007
8	Febrile seizures and generalized epilepsy associated with a mutation in the Na+-channel ÃŸ1 subunit gene SCN1B. Nature Genetics, 1998, 19, 366-370.	9.4	965
9	Mutations in filamin 1 Prevent Migration of Cerebral Cortical Neurons in Human Periventricular Heterotopia. Neuron, 1998, 21, 1315-1325.	3.8	811
10	Mutant GABAA receptor Î³2-subunit in childhood absence epilepsy and febrile seizures. Nature Genetics, 2001, 28, 49-52.	9.4	721
11	Prediction of seizure likelihood with a long-term, implanted seizure advisory system in patients with drug-resistant epilepsy: a first-in-man study. Lancet Neurology, The, 2013, 12, 563-571.	4.9	674
12	Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. Nature Genetics, 2013, 45, 825-830.	9.4	589
13	Epileptology of the first-seizure presentation: a clinical, electroencephalographic, and magnetic resonance imaging study of 300 consecutive patients. Lancet, The, 1998, 352, 1007-1011.	6.3	532
14	Autosomal dominant nocturnal frontal lobe epilepsy. Brain, 1995, 118, 61-73.	3.7	523
15	Temporal lobectomy: long-term seizure outcome, late recurrence and risks for seizure recurrence. Brain, 2004, 127, 2018-2030.	3.7	510
16	The spectrum of SCN1A-related infantile epileptic encephalopathies. Brain, 2007, 130, 843-852.	3.7	501
17	KCNQ2 encephalopathy: Emerging phenotype of a neonatal epileptic encephalopathy. Annals of Neurology, 2012, 71, 15-25.	2.8	427
18	Truncation of the GABAA-Receptor Î³2 Subunit in a Family with Generalized Epilepsy with Febrile Seizures Plus. American Journal of Human Genetics, 2002, 70, 530-536.	2.6	425

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19	Strikingly Different Clinicopathological Phenotypes Determined by Progranulin-Mutation Dosage. American Journal of Human Genetics, 2012, 90, 1102-1107.	2.6	414
20	X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. Nature Genetics, 2008, 40, 776-781.	9.4	397
21	An Insertion Mutation of the CHRNA4 Gene in a Family With Autosomal Dominant Nocturnal Frontal Lobe Epilepsy. Human Molecular Genetics, 1997, 6, 943-947.	1.4	381
22	Epilepsies in twins: Genetics of the major epilepsy syndromes. Annals of Neurology, 1998, 43, 435-445.	2.8	365
23	Hippocampal sclerosis in temporal lobe epilepsy demonstrated by magnetic resonance imaging. Annals of Neurology, 1991, 29, 175-182.	2.8	354
24	Paroxysmal exercise-induced dyskinesia and epilepsy is due to mutations in SLC2A1, encoding the glucose transporter GLUT1. Brain, 2008, 131, 1831-1844.	3.7	340
25	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
26	Missense mutations in the sodium-gated potassium channel gene KCNT1 cause severe autosomal dominant nocturnal frontal lobe epilepsy. Nature Genetics, 2012, 44, 1188-1190.	9.4	333
27	Sodium-channel defects in benign familial neonatal-infantile seizures. Lancet, The, 2002, 360, 851-852.	6.3	332
28	SCN1Amutations and epilepsy. Human Mutation, 2005, 25, 535-542.	1.1	327
29	GRIN2A mutations cause epilepsy-aphasia spectrum disorders. Nature Genetics, 2013, 45, 1073-1076.	9.4	326
30	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	13.9	326
31	Magnetic resonance imaging in temporal lobe epilepsy: Pathological correlations. Annals of Neurology, 1987, 22, 341-347.	2.8	324
32	Progressive Myoclonus Epilepsies: Specific Causes and Diagnosis. New England Journal of Medicine, 1986, 315, 296-305.	13.9	301
33	Mutations in DEPDC5 cause familial focal epilepsy with variable foci. Nature Genetics, 2013, 45, 546-551.	9.4	301
34	CHRNB2 Is the Second Acetylcholine Receptor Subunit Associated with Autosomal Dominant Nocturnal Frontal Lobe Epilepsy*. American Journal of Human Genetics, 2001, 68, 225-231.	2.6	300
35	GABRD encoding a protein for extra- or peri-synaptic GABAA receptors is a susceptibility locus for generalized epilepsies. Human Molecular Genetics, 2004, 13, 1315-1319.	1.4	299
36	De-novo mutations of the sodium channel gene SCN1A in alleged vaccine encephalopathy: a retrospective study. Lancet Neurology, The, 2006, 5, 488-492.	4.9	295

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37	Generalized epilepsy with febrile seizures plus: A common childhood-onset genetic epilepsy syndrome. Annals of Neurology, 1999, 45, 75-81.	2.8	271
38	Earlyâ€onset absence epilepsy caused by mutations in the glucose transporter GLUT1. Annals of Neurology, 2009, 66, 415-419.	2.8	266
39	Kufs Disease, the Major Adult Form of Neuronal Ceroid Lipofuscinosis, Caused by Mutations in CLN6. American Journal of Human Genetics, 2011, 88, 566-573.	2.6	253
40	Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurology, 2004, 55, 550-557.	2.8	250
41	Interictal spikes and epileptic seizures: their relationship and underlying rhythmicity. Brain, 2016, 139, 1066-1078.	3.7	250
42	Navigating the channels and beyond: unravelling the genetics of the epilepsies. Lancet Neurology, The, 2008, 7, 231-245.	4.9	249
43	KCNT1 gain of function in 2 epilepsy phenotypes is reversed by quinidine. Annals of Neurology, 2014, 75, 581-590.	2.8	249
44	SRPX2 mutations in disorders of language cortex and cognition. Human Molecular Genetics, 2006, 15, 1195-1207.	1.4	248
45	Title is missing!. Nature Genetics, 2001, 28, 49-52.	9.4	247
46	Placebo-controlled study of levetiracetam in idiopathic generalized epilepsy. Neurology, 2007, 69, 1751-1760.	1.5	246
47	A recurrent de novo mutation in KCNC1 causes progressive myoclonus epilepsy. Nature Genetics, 2015, 47, 39-46.	9.4	245
48	Definition and diagnostic criteria of sleep-related hypermotor epilepsy. Neurology, 2016, 86, 1834-1842.	1.5	245
49	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
50	Temporal lobe epilepsy and GEFS+ phenotypes associated with SCN1B mutations. Brain, 2006, 130, 100-109.	3.7	234
51	PRRT2 Mutations Cause Benign Familial Infantile Epilepsy and Infantile Convulsions with Choreoathetosis Syndrome. American Journal of Human Genetics, 2012, 90, 152-160.	2.6	234
52	The hidden genetics of epilepsyâ€”a clinically important new paradigm. Nature Reviews Neurology, 2014, 10, 283-292.	4.9	232
53	Phenotypic Characterization of an Î±₄ Neuronal Nicotinic Acetylcholine Receptor Subunit Knock-Out Mouse. Journal of Neuroscience, 2000, 20, 6431-6441.	1.7	231
54	Array-Based Gene Discovery with Three Unrelated Subjects Shows SCARB2/LIMP-2 Deficiency Causes Myoclonus Epilepsy and Glomerulosclerosis. American Journal of Human Genetics, 2008, 82, 673-684.	2.6	230

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55	GABRA1 and STXBP1 : Novel genetic causes of Dravet syndrome. Neurology, 2014, 82, 1245-1253.	1.5	229
56	Distinguishing Sleep Disorders From Seizures. Archives of Neurology, 2006, 63, 705.	4.9	223
57	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	2.8	222
58	Brivaracetam as adjunctive treatment for uncontrolled partial epilepsy in adults: A phase <scp>III</scp> randomized, doubleâ€blind, placeboâ€controlled trial. Epilepsia, 2014, 55, 57-66.	2.6	217
59	Exomeâ€based analysis of cardiac arrhythmia, respiratory control, and epilepsy genes in sudden unexpected death in epilepsy. Annals of Neurology, 2016, 79, 522-534.	2.8	216
60	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	3.7	215
61	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. Pharmacological Reviews, 2018, 70, 142-173.	7.1	215
62	Localization of epileptic foci with postictal single photon emission computed tomography. Annals of Neurology, 1989, 26, 660-668.	2.8	212
63	Familial temporal lobe epilepsy: A common disorder identified in twins. Annals of Neurology, 1996, 40, 227-235.	2.8	211
64	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	1.4	211
65	KUFS' DISEASE: A CRITICAL REAPPRAISAL. Brain, 1988, 111, 27-62.	3.7	210
66	A Homozygous Mutation in Human PRICKLE1 Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome. American Journal of Human Genetics, 2008, 83, 572-581.	2.6	199
67	NREM Arousal Parasomnias and Their Distinction from Nocturnal Frontal Lobe Epilepsy: A Video EEG Analysis. Sleep, 2009, 32, 1637-1644.	0.6	195
68	Mutations in mammalian target of rapamycin regulator DEPDC5 cause focal epilepsy with brain malformations. Annals of Neurology, 2014, 75, 782-787.	2.8	193
69	Reduced cortical inhibition in a mouse model of familial childhood absence epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17536-17541.	3.3	192
70	Human epilepsies: interaction of genetic and acquired factors. Trends in Neurosciences, 2006, 29, 391-397.	4.2	190
71	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	2.8	190
72	Extended spectrum of idiopathic generalized epilepsies associated with CACNA1H functional variants. Annals of Neurology, 2007, 62, 560-568.	2.8	186

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73	Mechanisms of human inherited epilepsies. Progress in Neurobiology, 2009, 87, 41-57.	2.8	185
74	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	2.8	184
75	Genetic Association Studies in Epilepsy: "The Truth Is Out There". Epilepsia, 2004, 45, 1429-1442.	2.6	179
76	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	2.8	175
77	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	4.9	175
78	Magnetic stimulation of the brain in generalized epilepsy: Reversal of cortical hyperexcitability by anticonvulsants. Annals of Neurology, 1993, 34, 351-355.	2.8	174
79	Mutations in the GABA Transporter SLC6A1 Cause Epilepsy with Myoclonic-Atonic Seizures. American Journal of Human Genetics, 2015, 96, 808-815.	2.6	173
80	Seizure-associated hippocampal volume loss: A longitudinal magnetic resonance study of temporal lobe epilepsy. Annals of Neurology, 2002, 51, 641-644.	2.8	172
81	Epilepsy and mental retardation limited to females: an under-recognized disorder. Brain, 2008, 131, 918-927.	3.7	172
82	Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Brain, 2013, 136, 3140-3150.	3.7	168
83	A Mutation in the Golgi Qb-SNARE Gene GOSR2 Causes Progressive Myoclonus Epilepsy with Early Ataxia. American Journal of Human Genetics, 2011, 88, 657-663.	2.6	166
84	A variant of <scp>KCC</scp> 2 from patients with febrile seizures impairs neuronal Cl ^{â˜} extrusion and dendritic spine formation. EMBO Reports, 2014, 15, 723-729.	2.0	163
85	Lateralization of verbal memory and unilateral hippocampal sclerosis: Evidence of task-specific effects. Neuropsychology, Development and Cognition Section A: Journal of Clinical and Experimental Neuropsychology, 1993, 15, 608-618.	1.4	159
86	Autosomal dominant rolandic epilepsy and speech dyspraxia: A new syndrome with anticipation. Annals of Neurology, 1995, 38, 633-642.	2.8	156
87	Hypothalamic Hamartoma and Seizures: A Treatable Epileptic Encephalopathy. Epilepsia, 2003, 44, 969-973.	2.6	153
88	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	1.8	153
89	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	2.6	152
90	Transcallosal Resection of Hypothalamic Hamartomas, with Control of Seizures, in Children with Gelastic Epilepsy. Neurosurgery, 2001, 48, 108-118.	0.6	150

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91	Optimizing genomic medicine in epilepsy through a gene-customized approach to missense variant interpretation. Genome Research, 2017, 27, 1715-1729.	2.4	150
92	Paroxysmal Motor Disorders of Sleep: The Clinical Spectrum and Differentiation from Epilepsy. Epilepsia, 2006, 47, 1775-1791.	2.6	149
93	Childhood absence epilepsy and febrile seizures: a family with a GABAA receptor mutation. Brain, 2003, 126, 230-240.	3.7	148
94	Occipital epilepsies: identification of specific and newly recognized syndromes. Brain, 2003, 126, 753-769.	3.7	142
95	Comparison of ictal SPECT and interictal PET in the presurgical evaluation of temporal lobe epilepsy. Annals of Neurology, 1995, 37, 738-745.	2.8	140
96	Risk factors for sudden unexpected death in epilepsy: a controlled prospective study based on coroners cases. Seizure: the Journal of the British Epilepsy Association, 2003, 12, 456-464.	0.9	140
97	Copy number variants are frequent in genetic generalized epilepsy with intellectual disability. Neurology, 2013, 81, 1507-1514.	1.5	140
98	Effects of angiotensin II on dopamine and serotonin turnover in the striatum of conscious rats. Brain Research, 1993, 613, 221-229.	1.1	137
99	Analyzing the Etiology of Benign Rolandic Epilepsy: A Multicenter Twin Collaboration. Epilepsia, 2006, 47, 550-555.	2.6	135
100	Genome-wide association analysis of genetic generalized epilepsies implicates susceptibility loci at 1q43, 2p16.1, 2q22.3 and 17q21.32. Human Molecular Genetics, 2012, 21, 5359-5372.	1.4	134
101	MR imaging and spectroscopic study of epileptogenic hypothalamic hamartomas: analysis of 72 cases. American Journal of Neuroradiology, 2004, 25, 450-62.	1.2	134
102	The genetics of human epilepsy. Trends in Pharmacological Sciences, 2003, 24, 428-433.	4.0	131
103	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.5	131
104	â€˜North Seaâ€™ progressive myoclonus epilepsy: phenotype of subjects with GOSR2 mutation. Brain, 2013, 136, 1146-1154.	3.7	129
105	Genetic Architecture of Idiopathic Generalized Epilepsy: Clinical Genetic Analysis of 55 Multiplex Families. Epilepsia, 2004, 45, 467-478.	2.6	128
106	Clinical challenges and future therapeutic approaches for neuronal ceroid lipofuscinosis. Lancet Neurology, The, 2019, 18, 107-116.	4.9	128
107	Mitochondrial dysfunction in multiple symmetrical lipomatosis. Annals of Neurology, 1991, 29, 566-569.	2.8	123
108	Glucose transporter 1 deficiency in the idiopathic generalized epilepsies. Annals of Neurology, 2012, 72, 807-815.	2.8	123

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109	Febrile seizures and hippocampal sclerosis: Frequent and related findings in intractable temporal lobe epilepsy of childhood. Pediatric Neurology, 1995, 12, 201-206.	1.0	122
110	Glucose Transporter 1 Deficiency as a Treatable Cause of Myoclonic Astatic Epilepsy. Archives of Neurology, 2011, 68, 1152.	4.9	121
111	Effects of vaccination on onset and outcome of Dravet syndrome: a retrospective study. Lancet Neurology, The, 2010, 9, 592-598.	4.9	119
112	The Genetics of Epilepsy. Annual Review of Genomics and Human Genetics, 2020, 21, 205-230.	2.5	116
113	Changes in cortical excitability differentiate generalized and focal epilepsy. Annals of Neurology, 2007, 61, 324-331.	2.8	114
114	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	3.7	112
115	Genetic epilepsy with febrile seizures plus. Neurology, 2017, 89, 1210-1219.	1.5	112
116	Familial partial epilepsy with variable foci: A new partial epilepsy syndrome with suggestion of linkage to chromosome 2. Annals of Neurology, 1998, 44, 890-899.	2.8	111
117	A Focal Epilepsy and Intellectual Disability Syndrome Is Due to a Mutation in TBC1D24. American Journal of Human Genetics, 2010, 87, 371-375.	2.6	111
118	Recent advances in the molecular genetics of epilepsy. Journal of Medical Genetics, 2013, 50, 271-279.	1.5	111
119	Validation of a Questionnaire for Clinical Seizure Diagnosis. Epilepsia, 1992, 33, 1065-1071.	2.6	110
120	Human Epileptogenesis and Hypothalamic Hamartomas: New Lessons from an Experiment of Nature. Epilepsia, 1997, 38, 1-3.	2.6	108
121	The peri-ictal state: cortical excitability changes within 24 h of a seizure. Brain, 2009, 132, 1013-1021.	3.7	108
122	Precision therapy for epilepsy due to KCNT1 mutations. Neurology, 2018, 90, e67-e72.	1.5	108
123	Sodium channels and the neurobiology of epilepsy. Epilepsia, 2012, 53, 1849-1859.	2.6	105
124	Cathepsin F mutations cause Type B Kufs disease, an adult-onset neuronal ceroid lipofuscinosis. Human Molecular Genetics, 2013, 22, 1417-1423.	1.4	105
125	Focal cortical myoclonus and rolandic cortical dysplasia: Clarification by magnetic resonance imaging. Annals of Neurology, 1988, 23, 317-325.	2.8	104
126	Altered kinetics and benzodiazepine sensitivity of a GABAA receptor subunit mutation [Â2(R43Q)] found in human epilepsy. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15170-15175.	3.3	104

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127	Parental Mosaicism in â€œDe Novoâ€•Epileptic Encephalopathies. New England Journal of Medicine, 2018, 378, 1646-1648.	13.9	104
128	Genetic variation ofCACNA1H in idiopathic generalized epilepsy. Annals of Neurology, 2004, 55, 595-596.	2.8	102
129	SCN2A Mutations and Benign Familial Neonatal-Infantile Seizures: The Phenotypic Spectrum. Epilepsia, 2007, 48, 1138-1142.	2.6	102
130	Channelopathies in idiopathic epilepsy. Neurotherapeutics, 2007, 4, 295-304.	2.1	101
131	Timing of De Novo Mutagenesis â€” A Twin Study of Sodium-Channel Mutations. New England Journal of Medicine, 2010, 363, 1335-1340.	13.9	100
132	Longâ€‰term followâ€‰up of febrile infectionâ€“related epilepsy syndrome. Epilepsia, 2012, 53, 101-110.	2.6	100
133	Clinical applications: MRI, SPECT, and PET. Magnetic Resonance Imaging, 1995, 13, 1119-1124.	1.0	99
134	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
135	Chromosomal Abnormalities and Epilepsy: A Review for Clinicians and Gene Hunters. Epilepsia, 2002, 43, 127-140.	2.6	98
136	Transcallosal Resection of Hypothalamic Hamartomas, with Control of Seizures, in Children with Gelastic Epilepsy. Neurosurgery, 2001, 48, 108-118.	0.6	97
137	Early onset absence epilepsy: 1 in 10 cases is caused by GLUT1 deficiency. Epilepsia, 2012, 53, e204-7.	2.6	97
138	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.5	97
139	Augmented currents of an HCN2 variant in patients with febrile seizure syndromes. Annals of Neurology, 2010, 67, 542-546.	2.8	96
140	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	2.8	96
141	Progressive Gait Deterioration in Adolescents With Dravet Syndrome. Archives of Neurology, 2012, 69, 873-8.	4.9	95
142	Familial cortical dysplasia type <scp>IIA</scp> caused by a germline mutation in <scp>DEPDC</scp>5. Annals of Clinical and Translational Neurology, 2015, 2, 575-580.	1.7	95
143	Limbic P3 potentials, seizure localization, and surgical pathology in temporal lobe epilepsy. Annals of Neurology, 1989, 26, 377-385.	2.8	94
144	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	2.8	94

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145	Familial neonatal seizures in 36 families: Clinical and genetic features correlate with outcome. Epilepsia, 2015, 56, 1071-1080.	2.6	94
146	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
147	Real-world utility of whole exome sequencing with targeted gene analysis for focal epilepsy. Epilepsy Research, 2017, 131, 1-8.	0.8	93
148	Susceptibility genes for complex epilepsy. Human Molecular Genetics, 2005, 14, R243-R249.	1.4	92
149	Genetics of the Epilepsies. Epilepsia, 2001, 42, 16-23.	2.6	91
150	Longâ€term seizure outcome and risk factors for recurrence after extratemporal epilepsy surgery. Epilepsia, 2012, 53, 970-978.	2.6	91
151	SCARB2 mutations in progressive myoclonus epilepsy (PME) without renal failure. Annals of Neurology, 2009, 66, 532-536.	2.8	90
152	Mapping of a Gene Determining Familial Partial Epilepsy with Variable Foci to Chromosome 22q11-q12. American Journal of Human Genetics, 1999, 65, 1698-1710.	2.6	89
153	Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. Brain, 2004, 127, 2173-2182.	3.7	89
154	The borderland of epilepsy: clinical and molecular features of phenomena that mimic epileptic seizures. Lancet Neurology, The, 2009, 8, 370-381.	4.9	88
155	Ictal 99mTc-HMPAO Single Photon Emission Computed Tomography in Children with Temporal Lobe Epilepsy. Epilepsia, 1993, 34, 869-877.	2.6	85
156	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	3.7	85
157	Predicting seizure control: Cortical excitability and antiepileptic medication. Annals of Neurology, 2010, 67, 64-73.	2.8	84
158	Channelopathies as a genetic cause of epilepsy. Current Opinion in Neurology, 2003, 16, 171-176.	1.8	82
159	Axon initial segment dysfunction in epilepsy. Journal of Physiology, 2010, 588, 1829-1840.	1.3	80
160	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.5	79
161	Comparison of Antiepileptic Drug Levels in Sudden Unexpected Deaths in Epilepsy with Deaths from Other Causes. Epilepsia, 1999, 40, 1795-1798.	2.6	78
162	Axon initial segment dysfunction in a mouse model of genetic epilepsy with febrile seizures plus. Journal of Clinical Investigation, 2010, 120, 2661-2671.	3.9	77

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163	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 706-712.	2.6	76
164	Benign mesial temporal lobe epilepsy. Nature Reviews Neurology, 2011, 7, 237-240.	4.9	76
165	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
166	PRRT2 phenotypic spectrum includes sporadic and fever-related infantile seizures. Neurology, 2012, 79, 2104-2108.	1.5	75
167	Epilepsy genetics: clinical impacts and biological insights. Lancet Neurology, The, 2020, 19, 93-100.	4.9	75
168	Mortality in patients with psychogenic nonepileptic seizures. Neurology, 2020, 95, e643-e652.	1.5	75
169	Epilepsy and mental retardation limited to females with PCDH19 mutations can present de novo or in single generation families. Journal of Medical Genetics, 2010, 47, 211-216.	1.5	74
170	Familial mesial temporal lobe epilepsy: a benign epilepsy syndrome showing complex inheritance. Brain, 2010, 133, 3221-3231.	3.7	74
171	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
172	Idiopathic Generalized Epilepsy with Generalized and Other Seizures inâ€ƒAdolescence. Epilepsia, 2002, 42, 317-320.	2.6	73
173	Hemicranial Volume Deficits in Patients with Temporal Lobe Epilepsy With and Without Hippocampal Sclerosis. Epilepsia, 1998, 39, 1174-1181.	2.6	72
174	Juvenile myoclonic epilepsy and idiopathic photosensitive occipital lobe epilepsy: is there overlap?. Brain, 2004, 127, 1878-1886.	3.7	72
175	Nicotine-Induced Dystonic Arousal Complex in a Mouse Line Harboring a Human Autosomal-Dominant Nocturnal Frontal Lobe Epilepsy Mutation. Journal of Neuroscience, 2007, 27, 10128-10142.	1.7	72
176	The psychological impact of a newly diagnosed seizure: Losing and restoring perceived control. Epilepsy and Behavior, 2007, 10, 223-233.	0.9	72
177	Genetic generalized epilepsies. Epilepsia, 2018, 59, 1148-1153.	2.6	72
178	Familial Partial Epilepsy with Variable Foci: Clinical Features and Linkage to Chromosome 22q12. Epilepsia, 2004, 45, 1054-1060.	2.6	71
179	Dominant KCNA2 mutation causes episodic ataxia and pharmacoresponsive epilepsy. Neurology, 2016, 87, 1975-1984.	1.5	71
180	Temporal Lobe Epilepsy Subtypes: Differential Patterns of Cerebral Perfusion on Ictal SPECT. Epilepsia, 1996, 37, 788-795.	2.6	70

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